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Thyroid sonographic abnormalities in McCune-Albright syndrome

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Abstract McCune-Albright syndrome (MAS) is characterised by the clinical triad precocious puberty, polyostotic bone dysplasia and café-au-lait skin lesions. Some studies have shown the possibility of multiple endocrinological disorders in this condition, especially thyroid disorders. We report the case of three girls with MAS and a heteromultinodular thyroid at sonography, despite the fact that they were clinically and biologically euthyroid. This raises the question of the follow-up and treatment of these lesions.

Introduction

McCune-Albright syndrome (MAS) is defined by the clinical triad of polyostotic dysplasia, café-au-lait pigmented skin lesions and precocious puberty. Additional endocrinological disorders have been described as part of this syndrome including hyperthyroidism, acromegaly, hyperparathyroidism and Cushing's syndrome [1, 2]. The most commonly reported of these involve the thyroid gland [3]. We report the case of three girls with MAS and abnormal thyroid sonograms.

Patients and methods

Three girls (aged 9, 13 and 19 years) with classic MAS were referred for follow-up of endocrine disorders. The disease was discovered in the patients at age 3.5, 5 and 4.5 years because of the classic triad (precocious puberty, café-au-lait skin lesions and fibrous dysplasia; Fig. 1 a). All three girls were clinically euthyroid at the time of evaluation and the thyroid gland was normal by palpation in two and enlarged in one. In the three cases, serum free T_4, T_3 and thyroid-stimulating hormone (TSH) levels were normal.

Thyroid ultrasonography (US) was systematically performed and abnormalities were found in the three patients. In patient 1, there was diffuse thyroid enlargement (lobe length: 50 mm). Multiple hypoechoic cyst-like areas (4–15 mm in diameter) were seen in both lobes (Fig. 1 b). Repeat studies after 6 months showed an increase in the size and pattern of some nodules. Because of the absence of uptake at scintigraphy, the left lobe and the isthmus were removed at operation, which also included a right superior parathyroidectomy, as palpation at surgery revealed an enlarged gland. The macroscopical analysis confirmed the presence of a heteromultinodular goitre, and the histological results showed multiple macrovesicular adenomas and parathyroid hyperplasia.

In patient 2, ultrasound indicated enlargement and diffuse inhomogeneity of the entire right lobe. There was one hypoechoic area at the lower pole suggestive of a cyst (12 mm in diameter), and multiple smaller hypoechoic regions were present throughout the remainder of the right lobe. The left thyroid lobe and isthmus were normal.

In patient 3, ultrasound confirmed the normal size of the gland and indicated that the entire left lobe had an inhomogeneous structure (Fig. 2). The right lobe was homogeneous but contained two small hypoechoic cyst-like regions (around 3 mm). In patients 2 and 3, no further investigations were performed due to the subclinical nature of the thyroid lesions.

Discussion

MAS is due to activating mutations of the α subunit of the G_s protein [4]. The G_s protein is responsible for signal transduction between receptor and adenylate cy-

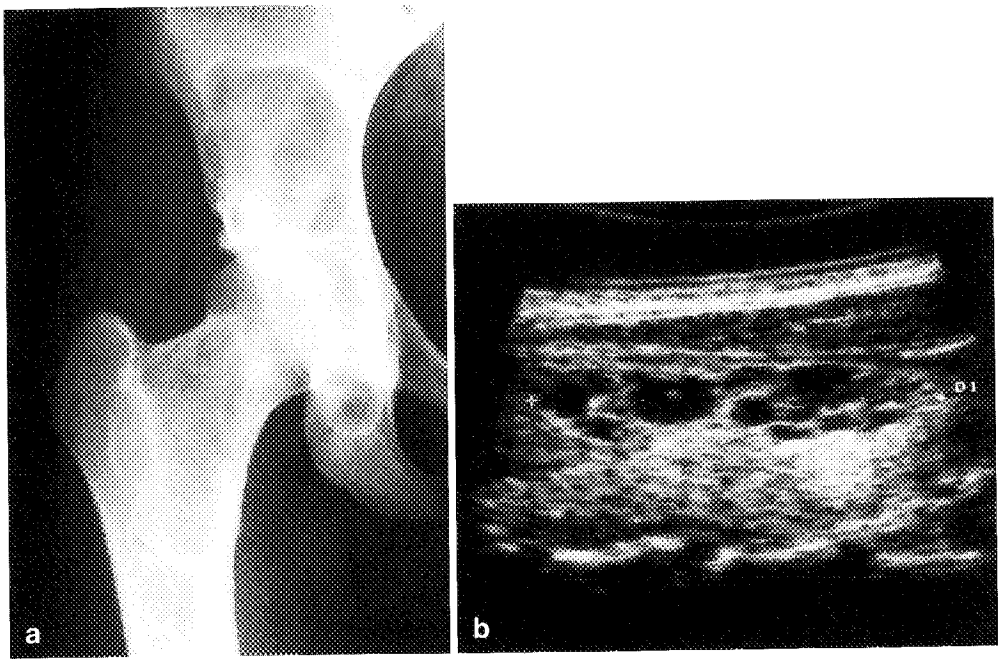

Fig. 1 **a** Patient 1. Plain film of the right hip showed typical fibrous dysplasia lesions. **b** Thyroid US: sagittal scan of the left lobe shows thyroid enlargement with multiple hypoechoic cyst-like areas. These were confirmed as macrovesicular adenomas by histological examination of the material obtained by resection of the left lobe and the isthmus

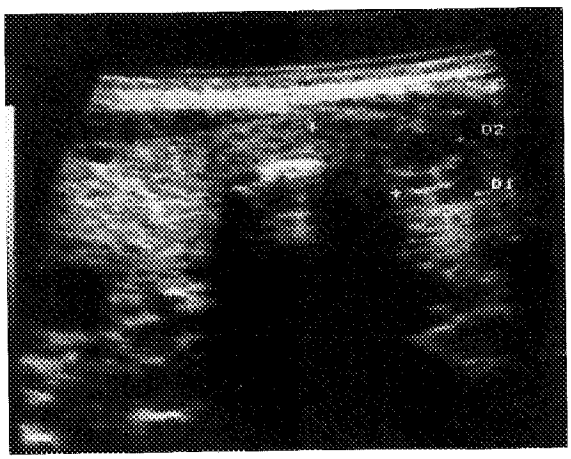

Fig. 2 Patient 2. Thyroid US: transverse scan shows an inhomogeneous structure in the left lobe and a small hypoechoic cyst-like lesion in the right lobe

clase, which is responsible for the synthesis of cyclic adenosine monophosphate (cAMP). This mutation occurs during the early stages of embryonic development, resulting in apparently random genetic mosaicism. Cells carrying the mutated G protein have a receptor-independent elevation of cAMP levels, resulting in constitutive activation. The apparently random distribution of mutated cells explains the diversity of clinical presentation of this syndrome.

Sexual precocity in MAS is due to ovarian hyperfunction despite low serum and urinary gonadotropin levels [2]. Resection of an ovarian cyst may lead to regression of secondary sexual characteristics. Similarly, hyperthyroidism in MAS is due to autonomous thyroid function, inducing suppression of TSH and a blunted response of TSH to thyrotropin-releasing hormone [5]. Thyroid ultrasonography was performed in three girls with MAS. These three girls were clinically euthyroid and they had normal serum thyroid hormone and TSH levels. Only one of the girls had thyroid enlargement detected by physical examination.

Thyroid ultrasonography appeared abnormal in all three cases but without any specific aspect, showing different abnormalities such as thyroid enlargement, inhomogeneous areas or hypoechoic cyst-like regions of various size. In one case, histological data confirmed the benignity of the thyroid lesions. Previous studies [2, 3, 5] have documented the occurrence of thyroid US abnormalities in MAS but these were associated with a thyroid dysfunction. In our study, none of the patients with US abnormalities showed clinical or biological hyperthyroidism. In one of these patients, a repeat study after 6 months showed an increase of the thyroid US abnormalities, whereas the bone disease remained stable. Of interest is that there was no apparent correlation between the severity of the bone disease and the presence or severity of the thyroid disorders in our patients.

The occurrence of US thyroid abnormalities in MAS raises the question of their follow-up and treatment, especially in the absence of endocrine dysfunction. Larger series are necessary to appreciate their exact frequency and histological nature. We suggest thyroid ultrasonography be performed routinely in patients with MAS.

References

1. Misaki M, Shima J, Ikoma J, Morioka K, Susuki S (1988) Acromegaly and hyperthyroidism associated with McCune-Albright syndrome. Horm Res 30: 26–28
2. Schwindinger WF, Levine MA (1993) McCune-Albright syndrome. Trends Endocrinol Metab 4: 238–242
3. Feuillan PP, Shawker T, Rose SR, Jones J, Jeevanram RK, Nisula BC (1990) Thyroid abnormalities in the McCune-Albright syndrome: ultrasonography and hormonal studies. J Clin Endocrinol Metab 71: 1596–1601
4. Weinstein LS, Shenker A, Gejman PV, Merino MJ, Friedman E, Spiegel AM (1991) Activating mutations of the stimulatory G protein in the McCune-Albright syndrome. N Engl J Med 325: 1688–1695
5. D'Armiento M, Reda G, Camagna A, Tardella L (1983) McCune-Albright syndrome: evidence for autonomous multiendocrine hyperfunction. J Pediatr 102: 584–586